Important News on Expanded Newborn Screening

As most of you are aware, SB 24 was passed in the 2005 Legislative Session to expand the number of conditions for which newborn screening is completed in Kentucky. The complete expansion will be effective December 31, 2005, and at that time a total of 29 conditions will be included on the screen which puts Kentucky in line with the national recommendation from the March of Dimes. Changes began in the newborn screening process effective July 2005 and the bullets below highlight important information for submitters of newborn screens and hospitals.

For All Submitters of Newborn Screens, including Hospitals:

- The new technology will detect disorders at 24 hours of age. The optimal specimen should be collected at 24 hours of age, but no later than 48 hours of age.
- If the infant is going to receive a blood transfusion, if possible, get the blood spot specimen prior to giving the transfusion even if the infant is not 24 hours of age.
- Antibiotics need to be documented on the filter paper card but will not automatically require a repeat.
- Demographic information and physician of record should be verified with the parent on the specimen to ensure that the physician and family can be contacted quickly in the situation of a positive screen.
- The specimen needs to be mailed to the state lab within 24 hours of collection, so the mail process at the submitting facility should examine their mailing procedure to assure entry into the USPS (United States Postal Service) as soon as possible after collection.
- There will be an educational presentation available on https://ky.train.org

Specific to Hospitals:

- Hospitals are required to have a newborn screening coordinator designated with the Department for Public Health Newborn Screening Program on an annual basis in January. Newborn Nursery nurse managers will be contacted to provide information.
- Hospitals will be required to implement a protocol to assure all newborns receive a newborn screening blood test and submit to the Department for Public Health.
- Hospitals will also be required to provide educational information to parents regarding newborn screening. This information is available on the HRSA website http://mchb.hrsa.gov/programs/default.htm and scroll down to Newborn Screening brochure.

Follow-Up:

Short Term follow-up for abnormal or unsatisfactory specimens is conducted by state staff at the Department for Public Health.

Abnormal Result

- The state lab notifies the follow-up staff of the abnormal result.
- The follow-up staff contacts the primary care physician listed on the NBS filter paper card by telephone with further action and faxes information to their office.
- The Department for Public Health contracts with University of Kentucky and the University of Louisville for specialty clinic referrals.

Unsatisfactory Specimen

- The laboratory staff mails out results either with a letter explaining to repeat only one test or if no letter is attached, the entire specimen needs to be repeated.
- If a repeat specimen is not received within 10 days, a letter is mailed to the parent explaining that no repeat has been received and to contact their baby's PCP.

After analyzing the data on the T4 levels the new cut off value will be 5.0ug/dL effective December 5, 2005. We will continue to monitor this and may adjust further in the future.

Disorders included in the screen as of December 31, 2005 are:

Disorders of Amino Acid Metabolism:

1. Phenylketonuria (PKU)
2. Maple Syrup Urine Disease (MSUD)
3. Homocystinuria (HCY)
4. Citrullinemia (CIT)
5. Arginosuccinic acidemia (ASA)
6. Tyrosinemia type 1 (TYR 1)

Disorders of Fatty Acid Oxidation

7. Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
8. Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
9. Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
10. Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
11. Trifunctional protein deficiency (TFP)
12. Carnitine uptake defect (CUD)

Disorders of Organic Acid Metabolism

13. Isovaleric acidemia (IVA)
14. Glutaric acidemia type 1 (GA 1)
15. 3-hydroxy-3-methyl glutaric aciduria (HMG)
16. Multiple carboxylase deficiency (MCD)
17. Methylmalonic acidemia (Cbl A, B)
18. Methylmalonic acidemia mutase deficiency (MUT)
19. Propionic Acidemia (PA)
20. β-ketothiolase deficiency (BKT)
21. 3-Methylcrotonyl-CoA carboxylase deficiency

Hemoglobinopathies

22. Sickle Cell Disease
23. Hemoglobin SC Disease
24. Hemoglobin S/β-thalassemia

Others

25. Galactosemia
26. Biotinidase deficiency
27. Congenital Adrenal Hyperplasia (CAH)
28. Cystic Fibrosis (CF)
29. Congenital Hypothyroidism (CH)

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